Chronic Granulomatous Disease

Chronic granulomatous disease: X-linked

(CGD; OMIM: 306400 (https://www.omim.org/entry/306400))

It is an X-linked primary immunodeficiency caused by a mutation of the **CYBB** gene (location Xp21.1), encoding the protein NOX 2 (previously referred to as glycoprotein gp91 *phox*). This is a part of the so-called cytochrome b -245 (also b 558), the catalytically active part of NADPH oxidase . NADPH-oxidase plays a key role in the respiratory burst of phages, as it produces superoxide [1][2]. If the formation of superoxide is impaired, other reactive forms of oxygen (hydrogen peroxide , hypochloric acid) are not formed eitheretc.), which serve as microbicidal agents of phages and participate in the degradation of absorbed particles. In addition, reactive oxygen species are also important for pH regulation in phagolysosomes, so their lack can also limit the activation of proteolytic phagosomal enzymes. The NADPH-oxidase defect thus causes a significant weakening of phagocytes , which have only a **limited ability** to dispose of phagocytosed material.

Reduced immunity is manifested from an early age **by purulent infections** . Increased sensitivity is especially to microorganisms producing catalase.

Therapy includes prophylactic administration of co-trimoxazole and itraconazole. In severe cases, a bone marrow transplant is considered .

Autosomally inherited forms

Chronic granulomatous disease: autosomal recessive Type 1

(CGD cytochrome b- positive 1; OMIM: 233700 (https://www.omim.org/entry/233700))

This autosomal recessive inherited form of chronic granulomatous disease is caused by a mutation of the **NCF1** gene (localization 7q11.23). The product of the gene is the NADPH oxidase subunit **p47-phox** . The respiratory burst of phages does not occur again. The clinical presentation is similar to that of the more common, X-linked form.

Chronic granulomatous disease: autosomal recessive Type 2

(CGD cytochrome b- positive 2; OMIM: 233710 (https://www.omim.org/entry/233710))

Another autosomal recessively inherited variant of chronic granulomatous disease is caused by a mutation in the gene for another NADPH-oxidase subunit, **p67-phox** (**NCF2** gene , 1q25 localization).

Links

Related articles

- Primary immunodeficiency
- Phagocytosis

Source

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. [cit. 21. 2. 2010]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Reference

1. GABRILOVICH, Dmitry I.. *the Neutrophils. New Outlook for Old Cells.* 2. edition. Imperial College Press, 2005. 355 pp. pp. 38-44. ISBN 1-86094-472-8.
2. VEJRAŽKA, Martin. *Úloha NAD(P)H oxidasy v signální transdukci [Doktorská dizertační práce].* 1. edition. Univerzita Karlova v Praze, 2007. 96 pp. pp. 36-38.

References

- BARTŮŇKOVÁ, Jiřina. *Imunodeficience.* 1. edition. Grada, 2002. 228 pp. ISBN 80-247-0244-4.